Effect Of Consanguineous Marriage On The Prevalence Of Recessive Trait On Muslim Population Of Saharsa District, Bihar (India). (Revised)

SYNOPSIS

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B. N. MANDAL UNIVERSITY, SINGHESHWAR,

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PROFESSOR & PRINCIPAL INVESTIGATOR
SURB-SURE DST PROJECT
DEPARTMENT OF ZOOLOGY
B.N. MANDAL UNIVERSITY, SINGHESHWAR,
MADHEPURA

RESEARCH SCHOLAR

ROLL NO. – ZOO-01/22

arshiakhan1404@gmail.com

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Introduction

Consanguineous marriage (Typically first-cousin and uncle–niece unions) increases homozygosity and the risk of autosomal recessive disorders, congenital anomalies, and early mortality [3] [4]. In India, consanguinity is common in some communities, and several studies document higher prevalence among Muslims and association with abortion, preterm delivery, and congenital anomalies compared to non-consanguineous unions.[5][2]. Muslims constitute about 14% of the total population of Saharsa district, providing a sizable community in which to study the genetic and health consequences of consanguinity in a relatively endogamous setting.[6][7].

Rationale and Research Questions

Rationale

There is limited district-level data from eastern India, particularly Bihar, linking measured consanguinity to specific recessive traits in Muslim populations, despite evidence from other Indian regions and neighboring countries that such unions increase offspring morbidity and mortality.[1][8]. Generating local evidence from Saharsa can support remarital counselling, community-based genetic education, and targeted screening for common recessive disorders.

Research questions

1. What is the prevalence and pattern (degree and type) of consanguineous marriage among Muslims in Saharsa district?
2. Whatistheprevalenceofselectedautosomalrecessivetraitsandcongenitalanomalies among offspring of consanguineous versus non-consanguineous Muslim couples?
3. Whatisthemagnitudeofassociationbetweenparentalconsanguinityandrecessivetraits after adjusting for socio-demographic and obstetric factors?

Objectives and Hypotheses

General Objective

To assess the effect of consanguineous marriage on the prevalence of autosomal recessive traits among the Muslim population of Saharsa district, Bihar.

Specific objectives

To estimate the proportion of Muslim marriages in Saharsa that are consanguineous and to classify them by degree (first cousin, second cousin, uncle–niece, etc.).

To document the occurrence of selected autosomal recessive conditions (e.g., congenital deafness, visual impairment, certain hemoglobin pathies, intellectual disability, and major congenital anomalies) among their offspring.

To compare the prevalence of the out comes between offspring of consanguineous and non-consanguineous couples and estimate relative risks/odds ratios.

To estimate the inbreeding coefficient at family level and explore its relationship with clinical outcomes.[9].

Hypotheses

H1: The prevalence of consanguineous marriage among Muslims in Saharsa is $\geq 20\%$, in line with data from Indian Muslim communities in other regions.[8][10].

H2: Offspring of consanguineous unions have significantly higher prevalence of selected autosomal recessive traits and congenital anomalies than off spring of non-consanguineous unions.

H3: Increasing degree of relatedness (higher inbreeding coefficient) is positively associated with the occurrence of recessive traits.

Study Setting and Design

Setting

Muslim households in selected rural and urban areas of Saharsa district, Bihar, where Muslims form a sizable minority with documented census data on population structure.[6][11].

Design

Community-based cross-sectional study with an embedded retrospective cohort component (reproductive and morbidity history of married couples), similar to designs used in prior Indian consanguinity and morbidity studies.[2][12].

Study Population and Sampling

Study population

Ever-married Muslim women (or couples) residing in Saharsa district with atleast one live birth, to allow assessment of offspring outcomes.[8]

Inclusion criteria

Muslim couples permanently residing in the selected clusters for ≥ 2 years. At least one biological child aged 0–18 years.

Exclusion criteria

Couples with adopted children only. Households unwilling to give informed consent.

Sampling strategy

Multi stage sampling:

Stage 1: Selection of blocks/urban wards with higher Muslim concentration using census

data. [6][13]

Stage 2: Random selection of villages/wards within these units.

Stage 3: Systematic sampling of eligible Muslim households.

Sample size (outline)

Calculate based on expected difference in prevalence of congenital anomalies or a key recessive trait between consanguineous and non-consanguineous unions, drawing effect-size estimates from Indian community studies where consanguineous marriages showed 40–60% prevalence and significantly higher morbidity.[5][2]

Variables and Operational Definitions

Exposure

Consanguineous marriage: Union between biologically related individuals upto second cousins, classified as: First-cousin (parallel, cross, patrilateral, matrilateral), Uncle–niece, Second-cousin or more distant, Non consanguineous marriage: Spouses unrelated upto second-cousin level. Degree of inbreeding quantified using standard inbreeding coefficient F for each union.[9].

Outcomes (recessive traits)

Documented autosomal recessive or strongly inbred-associated outcomes in offspring, such as: Congenital deafness and severe hearing impairment. Severe visual impairment and congenital eye anomalies. Major structural congenitalanomalies (cardiac, neural tube defects, limb anomalies, etc.). Clinically or laboratory diagnosed hemoglobin pathies (e.g., beta- thalassemia major, sickle cell disease where applicable).[3][12]. Intellectual disability or global developmental delay of presumed genetic origin.

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Covariates

Socio-demographic: parental age at marriage, education, occupation, socio-economic status, rural/urban residence.[8]. Reproductive history: parity,abortions, still births, preterm delivery, neonatal and infant deaths.[2][12]. Health-care access and consanguinity awareness variables in formed by previous awareness studies.[14][15].

Data Collection Methods

Tools

Pre-tested, structured interview schedule administered in local language to collect. Detailed pedigree upto three generations to classify degree of relatedness. Complete reproductive and morbidity history. Information on any diagnosed congenital disorders and chronic illness in offspring. Clinical examination of children by trained medical personnel and referral for confirmatory tests (audiometry, ophthalmologic exam, echocardiography, hemoglobin electrophoresis) where feasible, leveraging methods used in earlier Indian consanguinity and anomaly studies.[4][12]

Data sources

Household interviews with parents. Verification from medical records where available (hospital registers, immunization cards, disability certificates).

Data Analysis Plan

Descriptive statistics to estimate: Prevalence and types of consanguineous marriage among Muslims in Saharsa. Prevalence of each recessivetrait/congenital anomaly in offspring, stratified by consanguinity status.[2] Bivariate analysis (chi-square tests, crude odds ratios) to compare

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outcomes between consanguineous and non-consanguineous unions.[5][12]. Multivariable logistic regression to estimate adjusted odds ratios for key outcomes (e.g., any major congenital anomaly, any confirmed recessive disorder) controlling for confounders such as maternal age, parity, socioeconomic status, and rural/urban residence.[8][10]. Trend analysis for increasing inbreeding coefficient F versus risk of out comes, using F categories or continuous F.[9].

Ethical Considerations

Obtain ethical clearance from the institutional ethics committee and necessary permissions from local health authorities. Written informed consent from adult participants with assurances of confidentiality, anonymization of data, and voluntary participation. Sensitive handling of findings on genetic risk, with referral to appropriate counseling and clinical services; recommendations framed to respect cultural and religious practices while promoting awareness, as emphasized in Indian consanguinity awareness research.[16][15].

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Signature of Supervisor

Date: 01/12/25

Signature of Research Scholar

Date: 01.12-2025